

Diagnóstico genético y Medicina Personalizada

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Parque Científico de la Universidad de Valencia

www.imegen.es

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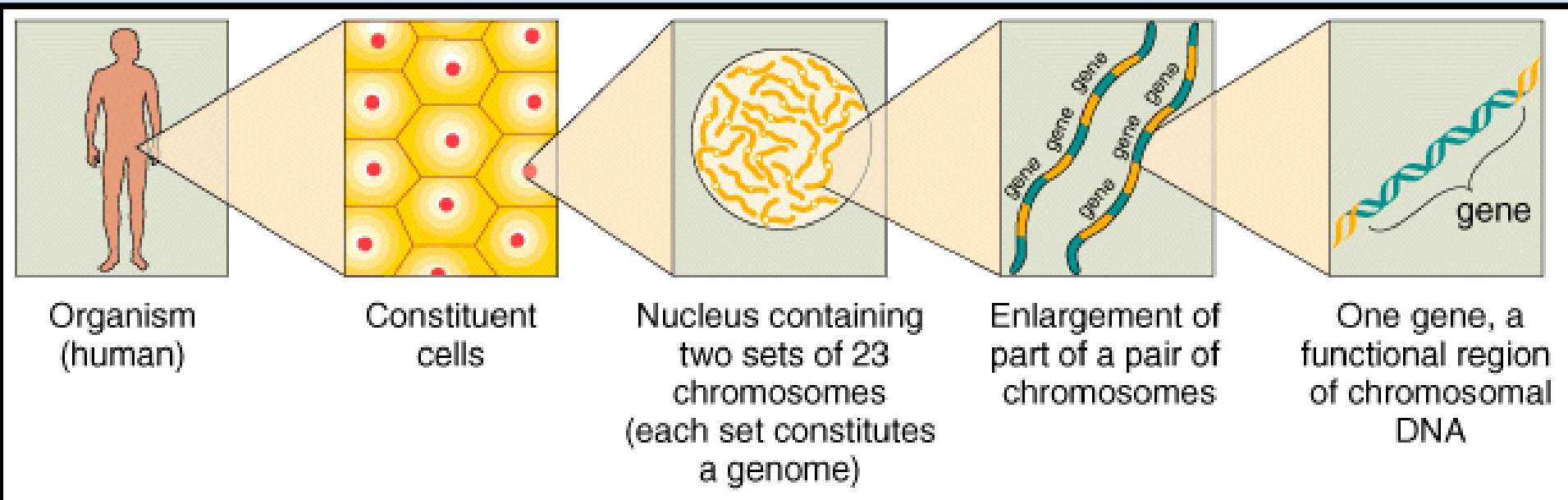
Los genes y el genoma

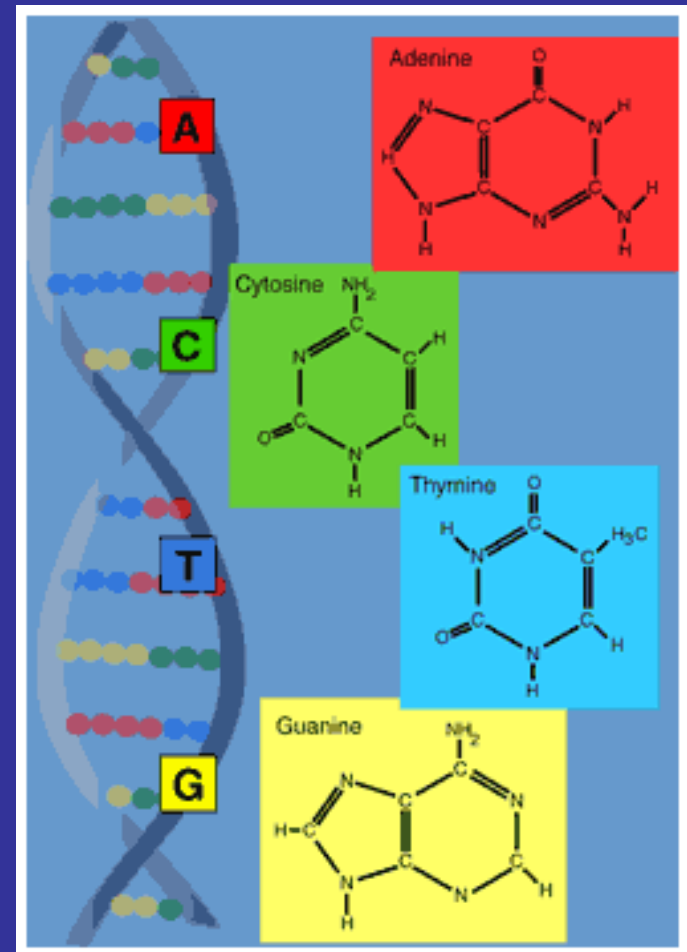
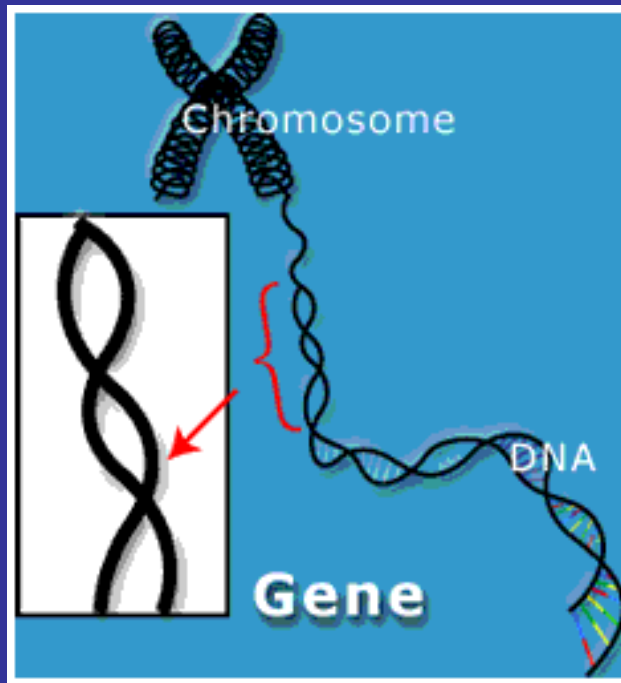


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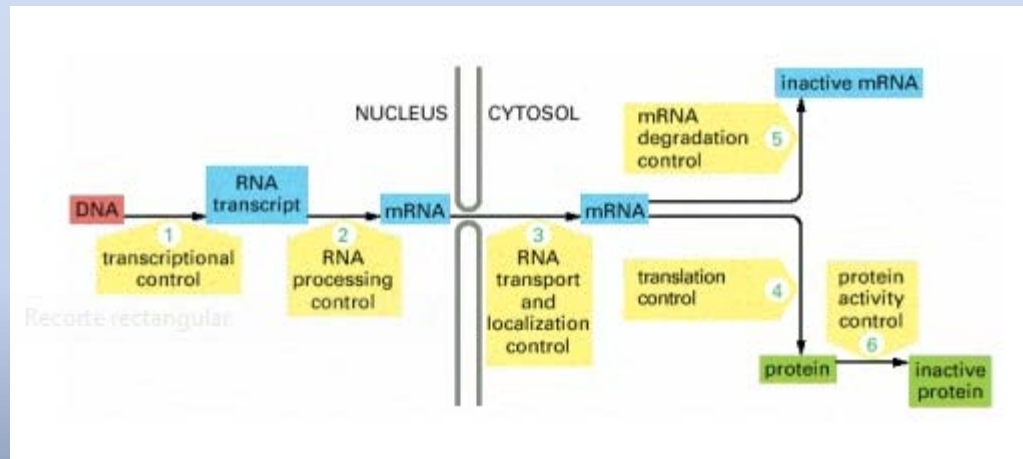
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Dos genomas en cada célula





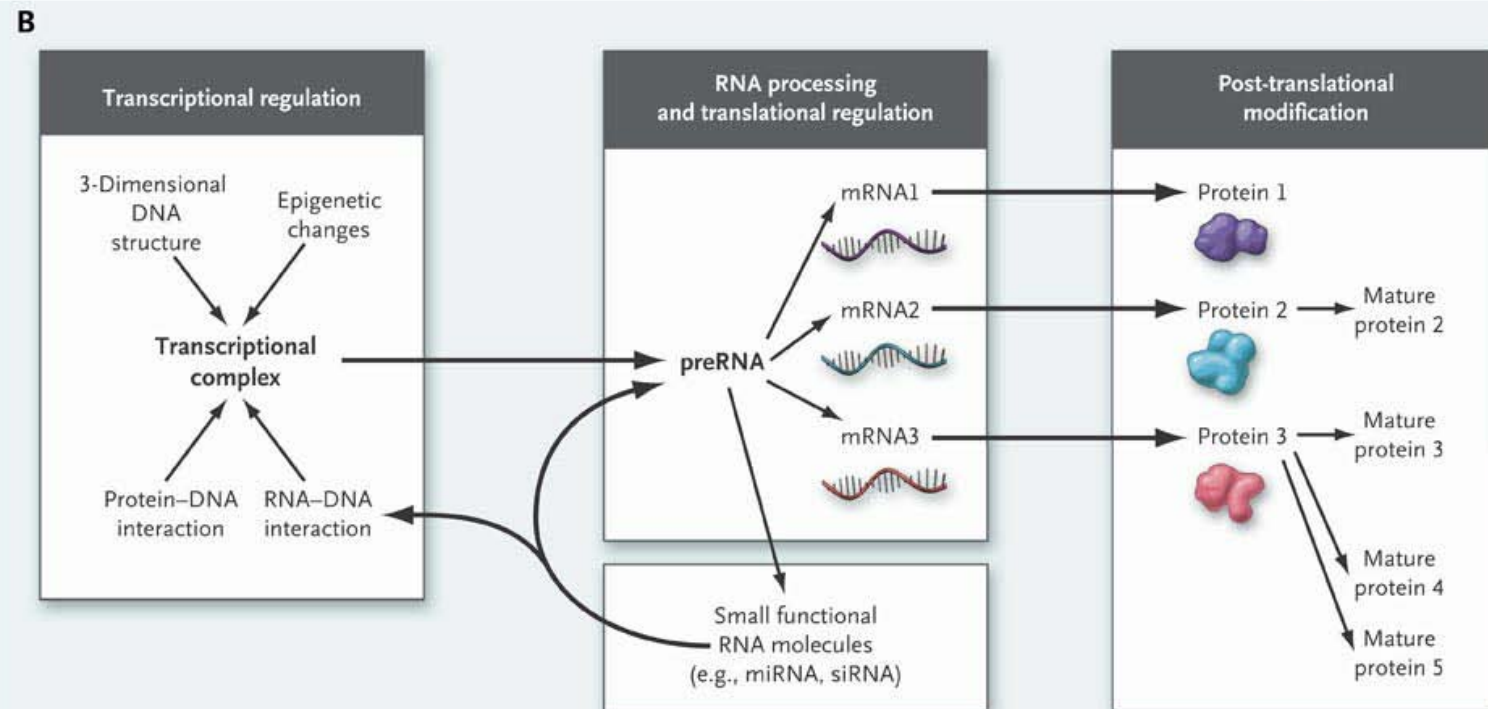
Dogma Central de la Biología Molecular



Copyright © 2002. Bruce Alberts, Alexander Johnson, Julian Lewis, Martin Raff, Keith Roberts, and Peter Walter. Molecular Biology of the Cell. Garland Science.



Dogma Central expandido



El genoma es una biblioteca



GAATTCGCCGGTTAGGTTTTAGTATCAAGAAGAGAAGGAAATTTTGGACAGTGAATGGGC
AATAAATGTGAGTAATATGGGAAATAAATGAATTTAAGAGGATTTGTTGTGATCGTTATT
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CATGGATTGTTTATTTGTTTATATCAATTTGTTTGCACCAACCACCACAAGAAAATTAGT
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ACTCATAACCTCTTCCTCATCCCCTTAAAAAACCTAAGAGTAGAGACTCTCTCAATCCC
GGCGGCGAAGATGAAGTACAACCCAAGAGTGACCTCTTCTCGCAGAAAGAACAGGAAGGC



Map Viewer

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NCBI Resources

[Genome Project](#)
[Trace Archive \(Venter\)](#)
[RefSeq](#)
[Whole Genome Association \(WGA\)](#)
[Human Genome Resources](#)
[GRC](#)

[Consensus CoDing Sequence \(CCDS\)](#)

[Trace FTP \(Personal Genomics\)](#)

[NCBI Handbook](#)

[Trace Archive \(Watson\)](#)

Organism Data in GenBank

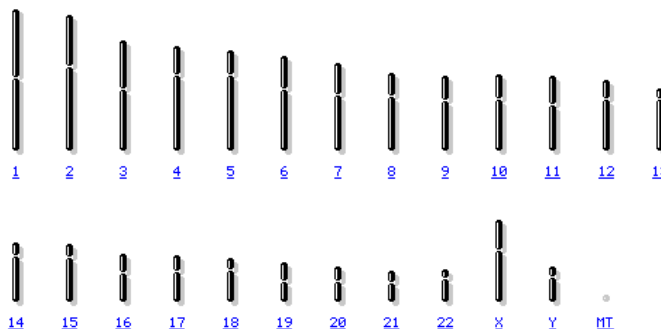
[EST](#)

[Genomic](#)

Homo sapiens (human) genome view

[Build 37.2 statistics](#) [Switch to previous build](#)

[BLAST search the human genome](#)



Lineage: [Eukaryota](#); [Metazoa](#); [Chordata](#); [Craniata](#); [Vertebrata](#); [Euteleostomi](#); [Mammalia](#); [Eutheria](#); [Euarchontoglires](#); [Primates](#); [Haplorrhini](#); [Catarrhini](#); [Hominidae](#); [Homo](#); [Homo sapiens](#)

November 2010: NCBI released an incremental update of the human genome reference assembly and updated annotation for all assemblies. The chromosomes and alternate loci regions are not changed; the new assembly includes the second set of genomic region Patches released by the Genome Reference Consortium ([Genome Reference Consortium \(GRC\)](#)) and is named GRCh37.p2. A previous version of the reference genome assembly, [NCBI Build 36.3](#), can still be accessed for Map Viewer display and for BLAST. For additional information about changes, statistics, and the status of the CCDS project please refer to:

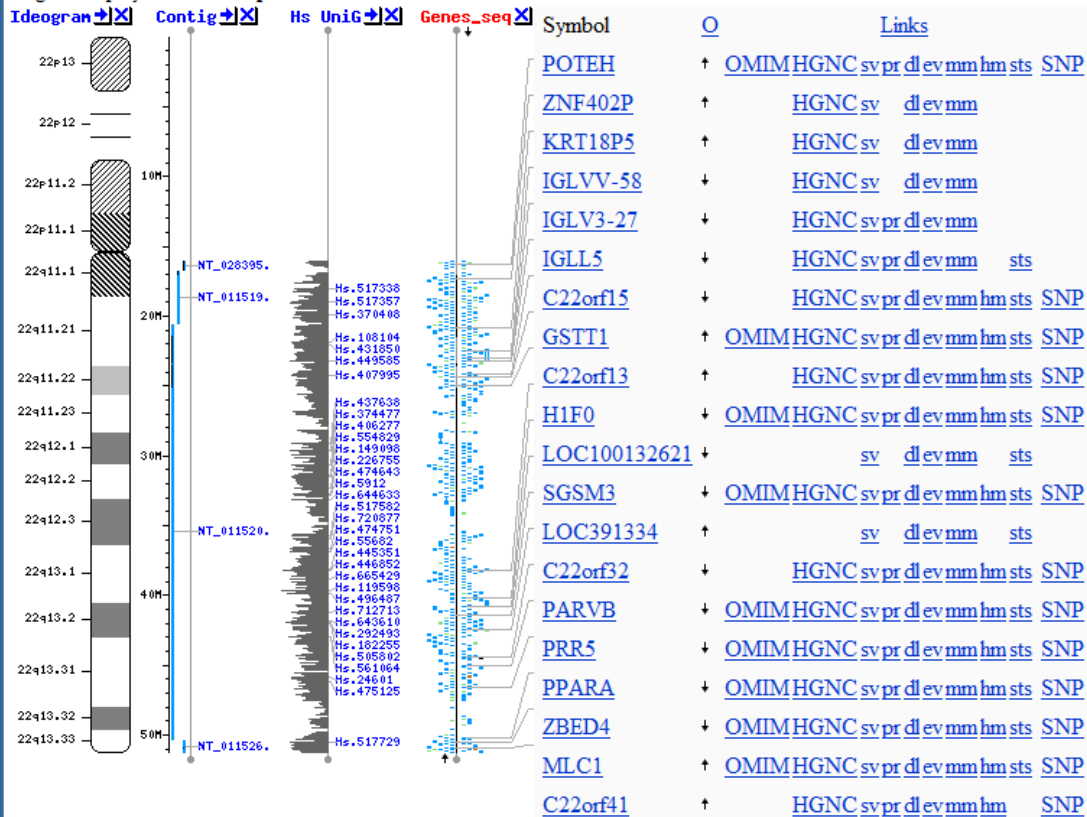
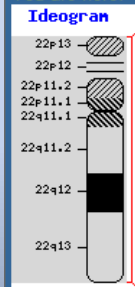
- [Release Notes](#)
- [Statistics](#)
- [CCDS Project](#)

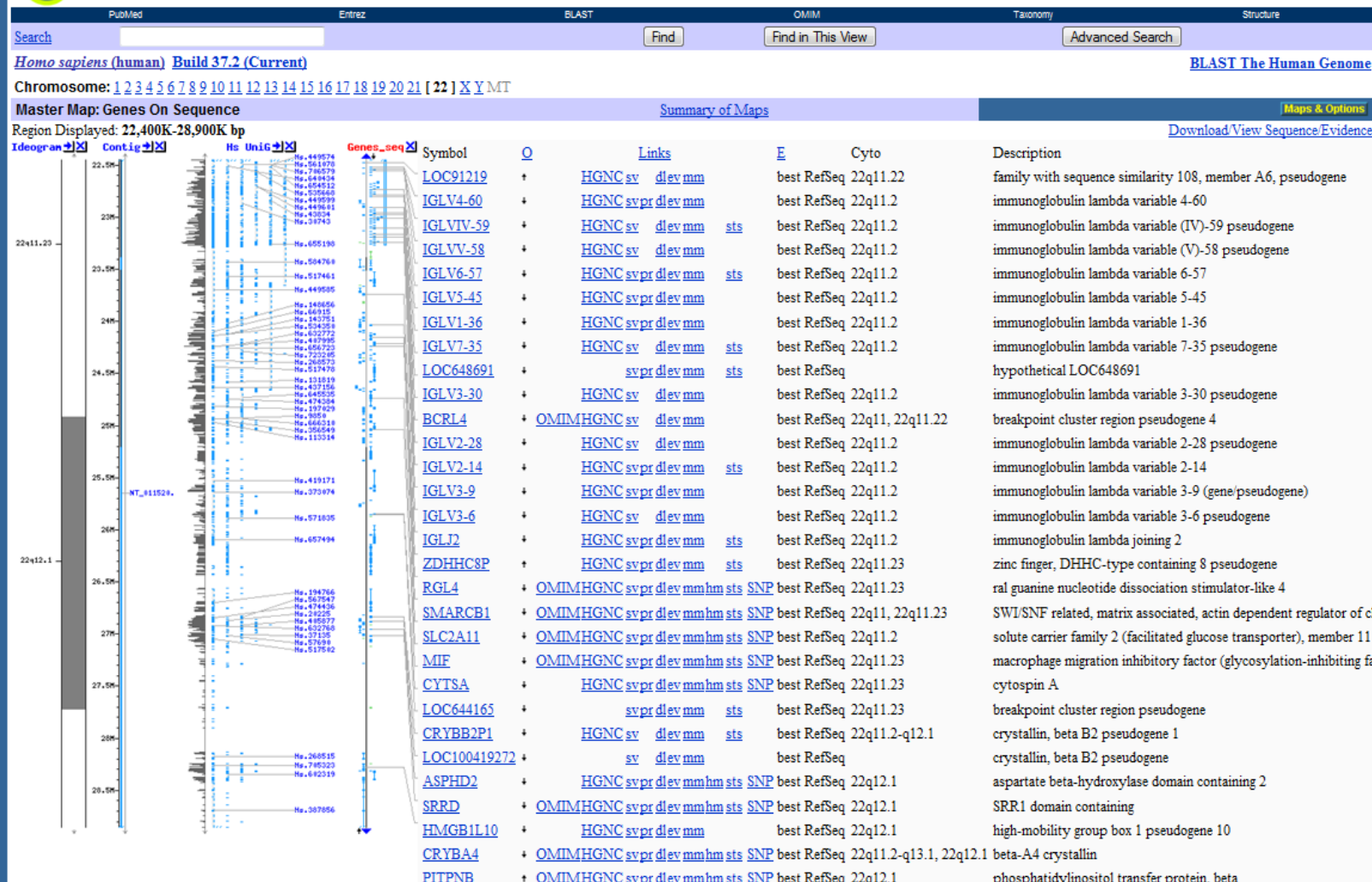
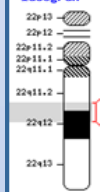
The NCBI Map Viewer provides graphical displays of features on the human genome sequence assembly as well as cytogenetic, genetic, physical, and radiation hybrid maps. Extensive [documentation](#) is provided to describe the resource features and methods used, tutorials, and statistics.

Map features that can be seen along the sequence include genes, transcripts, [NCBI contigs](#) (the 'Contig' map), the BAC tiling path (the 'Component' map), STSs, FISH mapped clones, ESTs and transcripts from several different organisms, [Gnomon](#) predicted gene models, and more.

www.ncbi.nlm.nih.gov/genome/guide/human







www.ncbi.nlm.nih.gov/genome/guide/human



El tamaño del genoma humano

3.000 millones de pb = 3 Gb

30.000 genes, aprox.



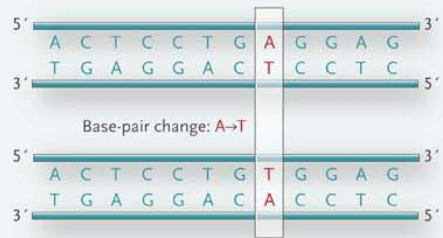
Las enfermedades genéticas



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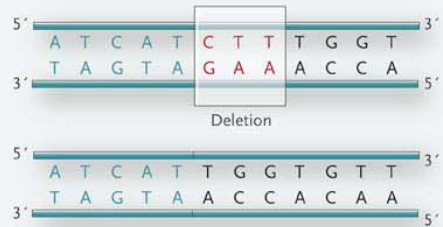
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imagen

A Single-base-pair changes

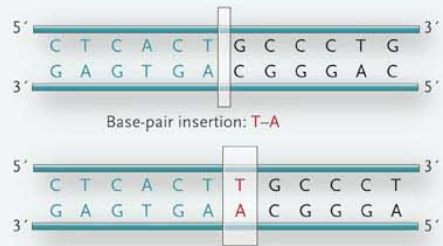


Example: **sickle cell disease**, A→T in human β -hemoglobin gene

B Insertions and deletions

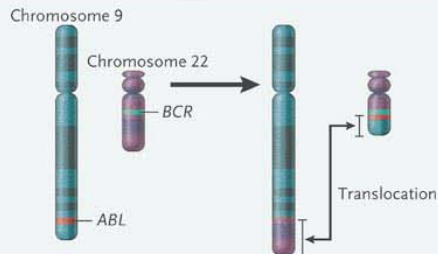


Example: **cystic fibrosis**, deletion of 3 base pairs, CTT, in the human *CFTR* gene



Example: **oculocutaneous albinism**, insertion of 1 base pair, T→A

C Structural rearrangements



Example: **chronic myelogenous leukemia**, chromosome 9 and 22 translocation, BCR-ABL gene fusion

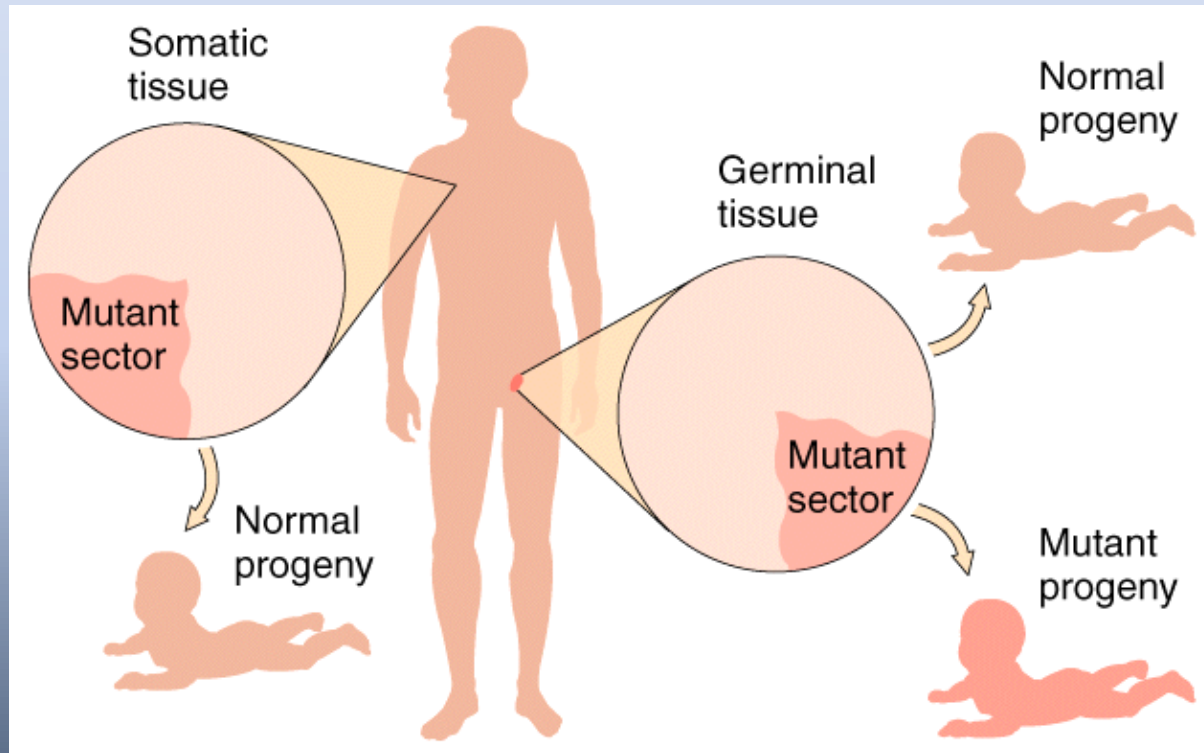
Mutaciones del genoma humano

Feero et al (2010). N Engl J Med 362: 2001-2011.



The NEW ENGLAND
JOURNAL of MEDICINE

Mutaciones heredables y mutaciones no heredables

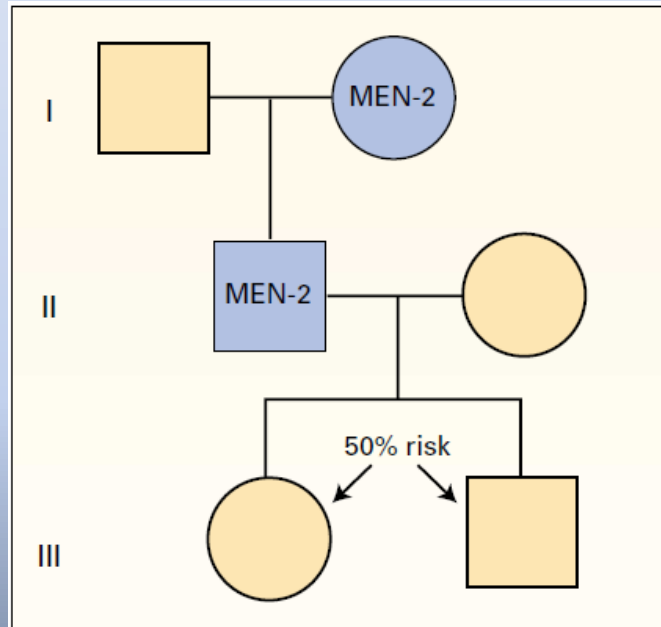


Dos tipos de herencia

- **Herencia mendeliana simple:** son enfermedades monogénicas (casi siempre enfermedades raras).
- **Herencia compleja:** son enfermedades multifactoriales (casi siempre enfermedades comunes, aunque algunas son enfermedades raras).



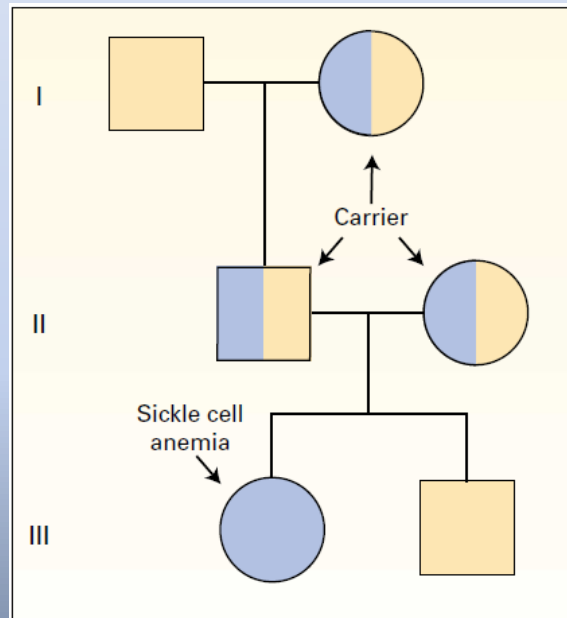
Herencia autosómica dominante



(Ej. Neoplasia endocrina múltiple)



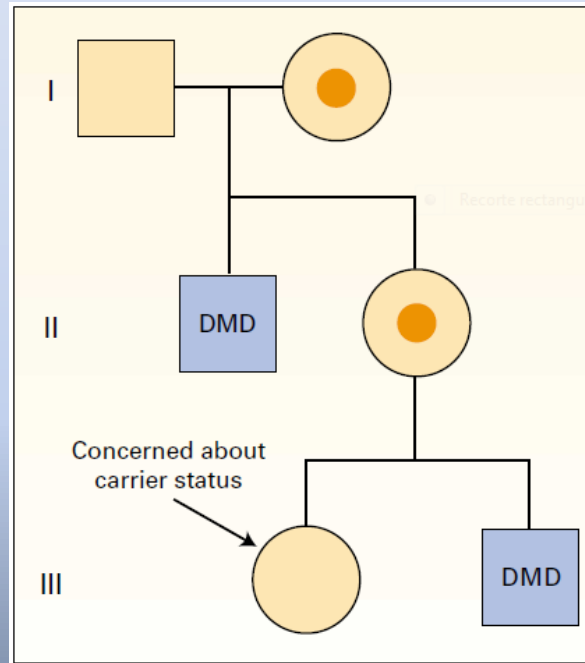
Herencia autosómica recesiva



(Ej. Anemia falciforme)



Herencia recesiva ligada al X



(Ej. Distrofia muscular de Duchenne)



Ejemplos de enfermedades genéticas

Fibrosis quística

Hemofilia

Síndrome de Marfan

Talasemia

Poliquistosis renal

Distrofia miotónica

Retinitis pigmentosa

Ataxias hereditarias

Síndrome de Prader-Willi

Cáncer de mama y ovario

Osteogénesis imperfecta

Distrofia muscular de Duchenne

Enfermedad de Huntington

Neurofibromatosis

Enfermedad de Charcot-Marie-Tooth

Fenilcetonuria

Síndrome del X-frágil

Acondroplasia (enanismo)

Hemocromatosis

Hipercolesterolemia familiar

Neoplasia endocrina múltiple

Enfermedad de Alzheimer



**Cerca de 6.000 enfermedades
genéticas, de las cuales ya podemos
diagnosticar unas 2.000,
(analizando genes individuales)**



All Databases PubMed Nucleotide Protein Genome Structure PMC OMIM

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[All: 1](#) [OMIM UniSTS: 0](#) [OMIM dbSNP: 0](#)

MIM ID +113705

[MGI](#), [GeneTests](#), [Links](#)

BREAST CANCER 1 GENE; BRCA1

Other entities represented by this entry

PANCREATIC CANCER, SUSCEPTIBILITY TO, 4, INCLUDED; PNCA4, INCLUDED

Gene map locus: [17q21](#)

Description

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BRCA1 plays critical roles in DNA repair, cell cycle checkpoint control, and maintenance of genomic stability. BRCA1 forms several distinct complexes through association with different adaptor proteins, and each complex forms in a mutually exclusive manner (Wang et al., 2009).

Cloning

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Miki et al. (1994) identified cDNA sequences corresponding to the BRCA1 gene by positional cloning of the region on 17q21 implicated in familial breast-ovarian cancer syndrome (604370). The deduced 1,863-residue protein with zinc-finger domains near the N terminus. A 7.8-kb mRNA transcript was identified in testes, thymus, breast and ovary. There appeared to be a complex pattern of alternative splicing.

Bennett et al. (1995) found that the mouse Brca1 gene shares 75% identity of the coding region with the human sequence at the nucleotide level, whereas the predicted amino acid identity was only 58%.

Jensen et al. (1996) demonstrated that BRCA1 encodes a 190-kD protein with sequence homology and biochemical analogy to members of the granin protein family, including chromogranin A (118910), chromogranin B (118920), and secretogranin II, also known as chromogranin C (118930). They noted that BRCA2 also includes a motif similar to the granin consensus at the C terminus of the protein. Both BRCA1 and the granins localize to secretory vesicles, are secreted by a regulated pathway, are posttranslationally glycosylated, and are responsive to hormones. The authors stated that as a regulated secretory protein, BRCA1 appears to function by a mechanism not previously described for tumor suppressor products. As reviewed by Steeg (1996), granins are a family of acidic proteins that bind calcium and aggregate in its presence. Known members of the granin family have been solely neuroendocrine or endocrine in origin; if BRCA1 is a granin it will necessarily expand the protein family boundaries.

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Links

Selected Gene Related Links
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OMIM Statistics for November 8, 2010

Number of Entries

	Autosomal	X-Linked	Y-Linked	Mitochondrial	Total
* Gene with known sequence	12526	615	48	35	13224
+ Gene with known sequence and phenotype	345	19	0	2	366
# Phenotype description, molecular basis known	2627	233	4	28	2892
% Mendelian phenotype or locus, molecular basis unknown	1634	133	5	0	1772
Other, mainly phenotypes with suspected mendelian basis	1847	130	2	0	1979
Total	18979	1130	59	65	20233

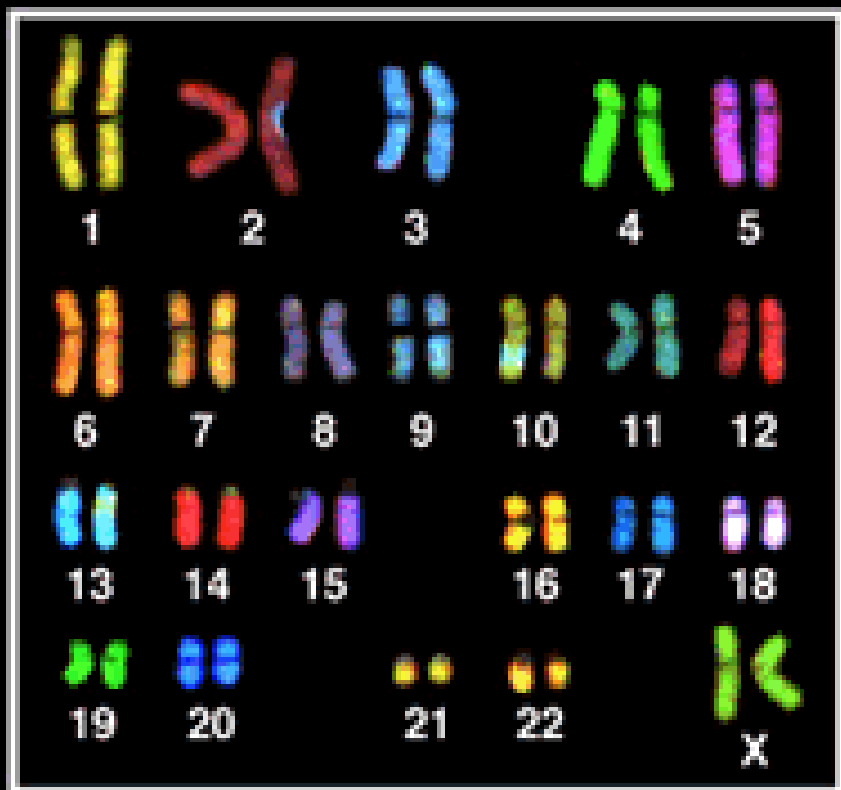
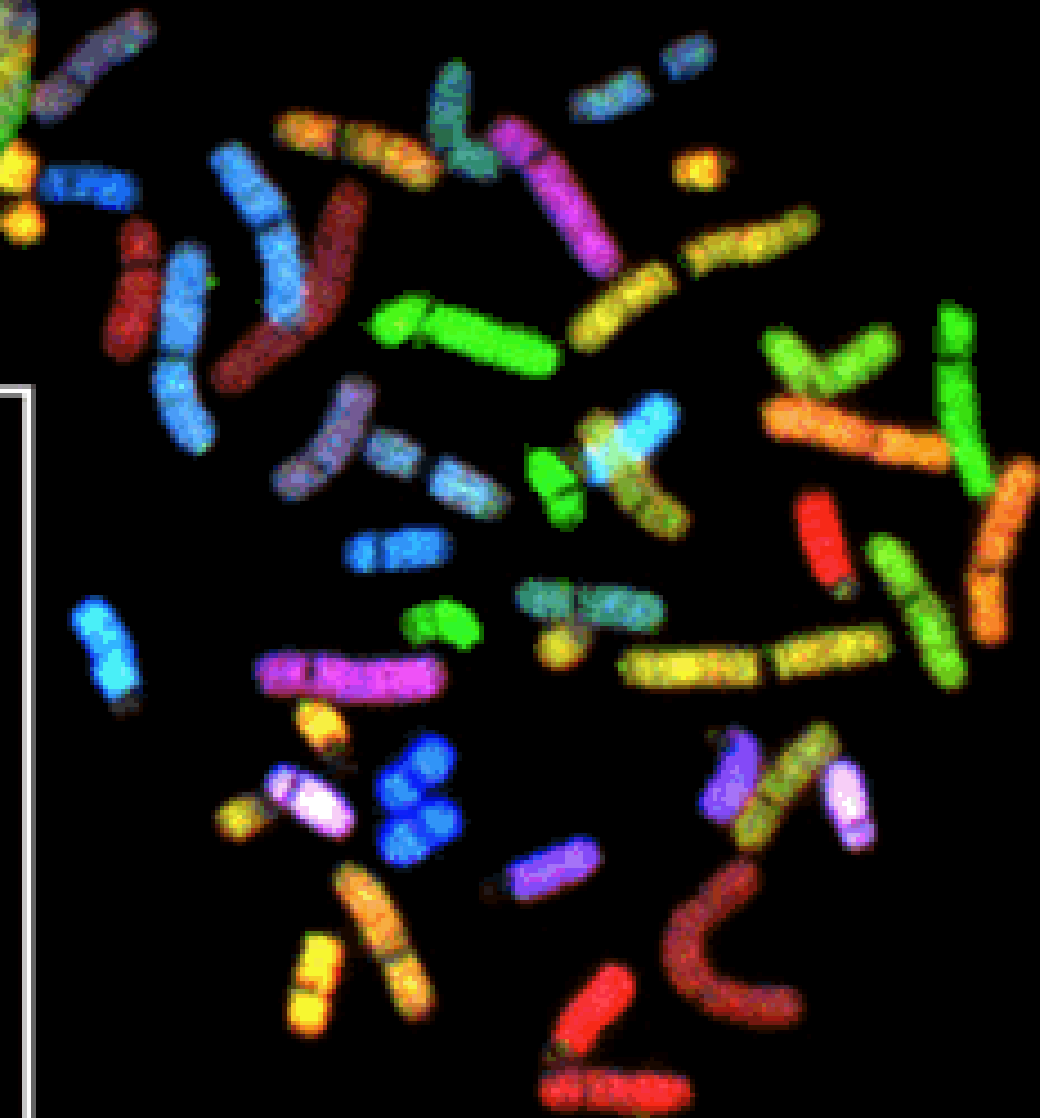
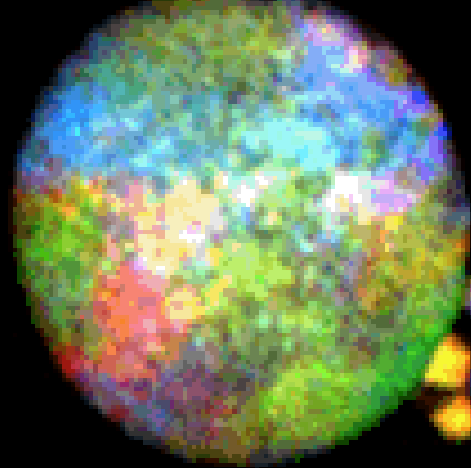


El diagnóstico genético

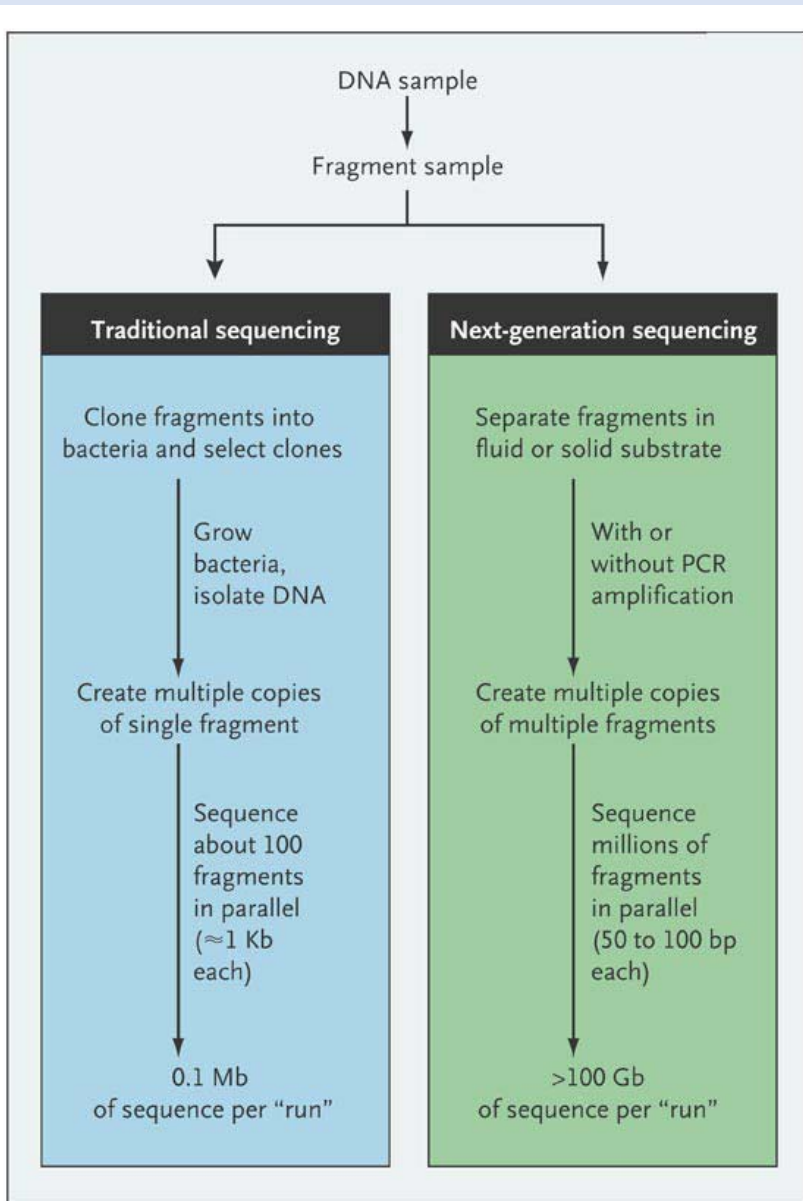


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Técnicas para la secuenciación de ADN



Secuenciación capilar (método de Sanger) VS Ultrasecuenciación

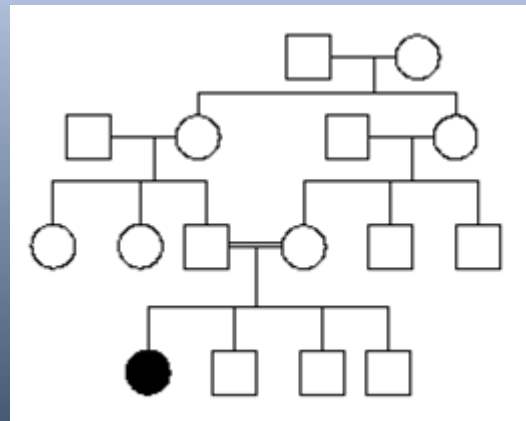
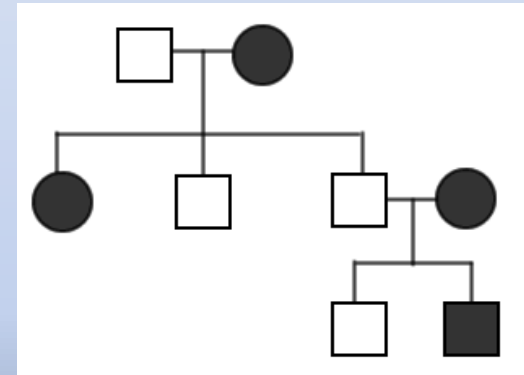
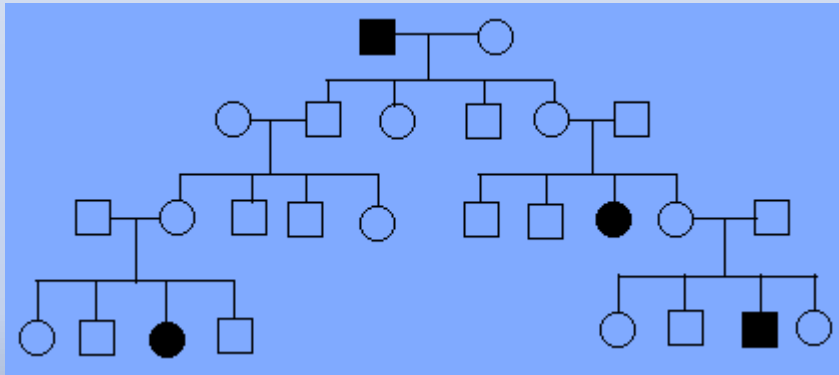
Feero et al (2010). N Engl J Med 362: 2001-2011.

El proceso del diagnóstico genético

1. Consulta del paciente a su médico: valoración de antecedentes familiares.
2. Prescripción del análisis: toma de muestra.
3. Envío de la muestra al Laboratorio de Referencia.
4. Extracción de ADN genómico a partir de sangre del paciente.
5. Acceso a la región genómica relevante mediante amplificación por PCR.
6. Secuenciación del gen (o genes) relevante.
7. Comparación de la secuencia de ADN del paciente con las secuencias referencia y análisis bioinformático.
8. Comunicación de resultados.
9. Consejo genético antes y después del test .

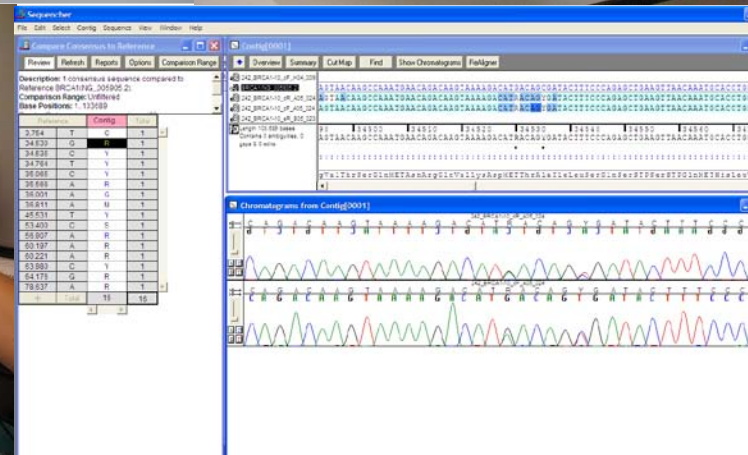
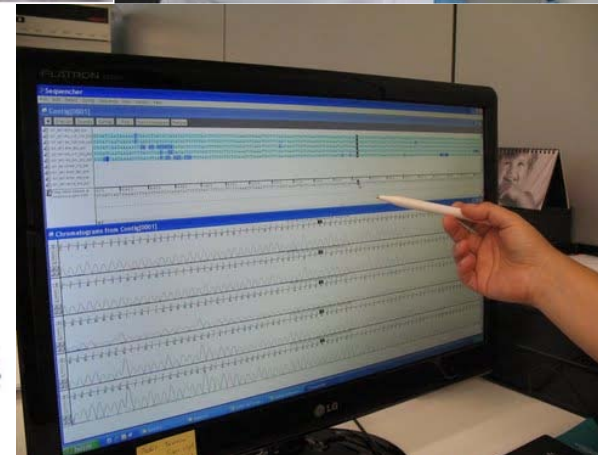
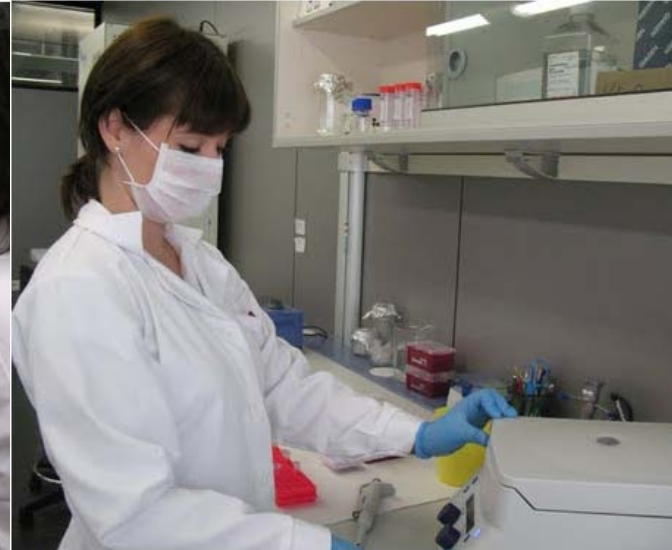


Estudio de antecedentes familiares: árbol genealógico



Acceso a la región relevante del genoma: PCR





Extracción y análisis del ADN





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Locus-specific databases

Other useful links



[HGMD Professional](#) includes 1. Up-to-date mutation data; 2. Fulltext indexing; 3. Advanced search facility; 4. Downloadable results; 5. And much more....! See the many [benefits](#) of HGMD Professional, take the [tour](#) (YouTube) and see the [benefits](#) of HGMD Professional.

This database is maintained by D.N. Cooper, E.V. Ball, P.D. Stenson, A.D. Phillips, K. Howells and M.E. Mort with the assistance of N.S.T. Thomas.



*Please note that this less up-to-date public version of our database is freely available only to [registered](#) users from academic institutions/non-profit organisations. All commercial users are required to purchase a license from BIOBASE, our commercial partner. A license to [HGMD Professional](#) is available to both commercial and academic/non-profit users wishing to access the most up-to-date version of the database (see [example](#) HGMD Professional entry). Read more about how HGMD is [funded](#).

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Table:	Description:	Public entries: <small>This site. Academic non-profit users only</small>	Total entries: <small>HGMD Professional 2010.3</small>
Mutation totals (as of 2010-11-10)		76676	105135
Gene symbol	The gene description, gene symbol (as recommended by the HUGO Nomenclature Committee) and chromosomal location is recorded for each gene. In cases where a gene symbol has not yet been made official, a provisional symbol has been adopted which is denoted by lower-case letters.	2911	3889
cDNA sequence	cDNA sequences are presented numbered by codon.		
Missense/nonsense	Single base-pair substitutions in coding regions are presented in terms of a triplet change with an additional flanking base included if the mutated base lies in either the first or third position in the triplet.	43639	58984
Splicing	Mutations with consequences for mRNA splicing are presented in brief with information specifying the relative position of the lesion with respect to a numbered intron donor or acceptor splice site. Positions given as positive integers refer to a 3' (downstream) location, negative integers refer to a 5' (upstream) location.	7347	9969
Regulatory	Substitutions causing regulatory abnormalities are logged in with thirty nucleotides flanking the site of the mutation on both sides. The location of the mutation relative to the transcriptional initiation site, initiation codon, polyadenylation site or termination codon is given.	1141	1873
Small deletions	Micro-deletions (20 bp or less) are presented in terms of the deleted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the caret character (^).	12358	16497
Small insertions	Micro-insertions (20 bp or less) are presented in terms of the inserted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the	5016	6802

Comparación frente a bases de datos de mutaciones





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MutaDATABASE is a publicly available, open access, free, online database that provides standardised information on human disease genes, variations, and clinical features of patients with monogenic disease.

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New genes in MutaDATABASE	
New gene reviews in MutaREVIEWS	
Total number of genes in MutaDATABASE	
Total number of gene reviews in MutaREVIEWS	

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News

Announcing new collaboration

Sept 10, 2010:

Announcing collaboration with The Human Variome Project

The Human Variome Project (HVP, represented by Dick Cotton), the LOVD (Leiden Open Variation Database, represented by Johan Den Dunnen) are joining the MutaDATABASE project as the 3 parties are each engaged in complementary activities to (a) promote the collection of and/or (b) collect data of genetic variants that cause inherited disease or genetic traits. The three groups now have come to an agreement to join activities to enhance the rate or progress of collection of the above-mentioned data so it can be used in testing, counseling, therapy, research or inherited disorders.

The 3 parties will systematically and regularly dump data from their locus-specific databases into each other's locus-specific databases so that submission of data to any of these 3 databases will result in listing the data in all 3 mutation databases.

Iniciativas para unificar las bases de datos de mutaciones



La Medicina Genómica y Medicina Personalizada



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Del análisis de genes individuales al análisis del genoma completo





Perspective

JULY 22, 2010

The Path to Personalized Medicine

Margaret A. Hamburg, M.D., and Francis S. Collins, M.D., Ph.D.

Major investments in basic science have created an opportunity for significant progress in clinical medicine. Researchers have discovered hundreds of genes that harbor variations contributing

to human illness, identified genetic variability in patients' responses to dozens of treatments, and begun to target the molecular causes of some diseases. In addition, scientists are developing and using diagnostic tests based on genetics or other molecular mechanisms to better predict patients' responses to targeted therapy.

The challenge is to deliver the benefits of this work to patients. As the leaders of the National Institutes of Health (NIH) and the Food and Drug Administration (FDA), we have a shared vision of personalized medicine and the scientific and regulatory structure needed to support its growth. Together, we have been

focusing on the best ways to develop new therapies and optimize prescribing by steering patients to the right drug at the right dose at the right time.

We recognize that myriad obstacles must be overcome to achieve these goals. These include scientific challenges, such as determining which genetic markers have the most clinical significance, limiting the off-target effects of gene-based therapies, and conducting clinical studies to identify genetic variants that are correlated with a drug response. There are also policy challenges, such as finding a level of regulation for genetic tests that both protects patients and encourages innovation. To make progress,

the NIH and the FDA will invest in advancing translational and regulatory science, better define regulatory pathways for coordinated approval of codeveloped diagnostics and therapeutics, develop risk-based approaches for appropriate review of diagnostics to more accurately assess their validity and clinical utility, and make information about tests readily available.

Moving from concept to clinical use requires basic, translational, and regulatory science. On the basic-science front, studies are identifying many genetic variations underlying the risks of both rare and common diseases. These newly discovered genes, proteins, and pathways can represent powerful new drug targets, but currently there is insufficient evidence of a downstream market to entice the private sector to explore most of them. To fill that void, the NIH and the FDA will

The new science of personalized medicine

The new science of personalized medicine: Translating the promise into practice



PMC Personalized
Medicine Coalition

The Case for

PERSONALIZED MEDICINE

*We shed light on the demonstrated benefits
of personalized medicine and describe the
pathway for its widespread adoption to
improve healthcare.*



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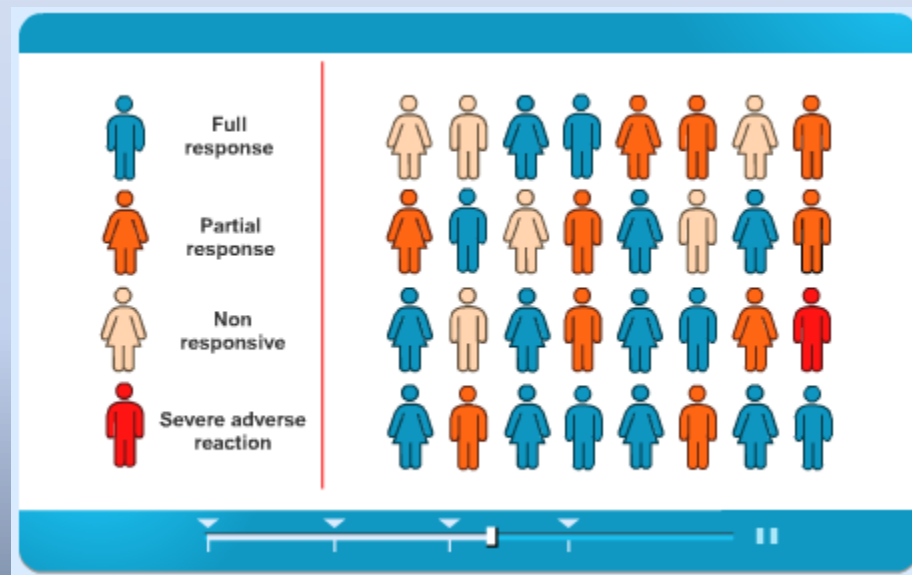
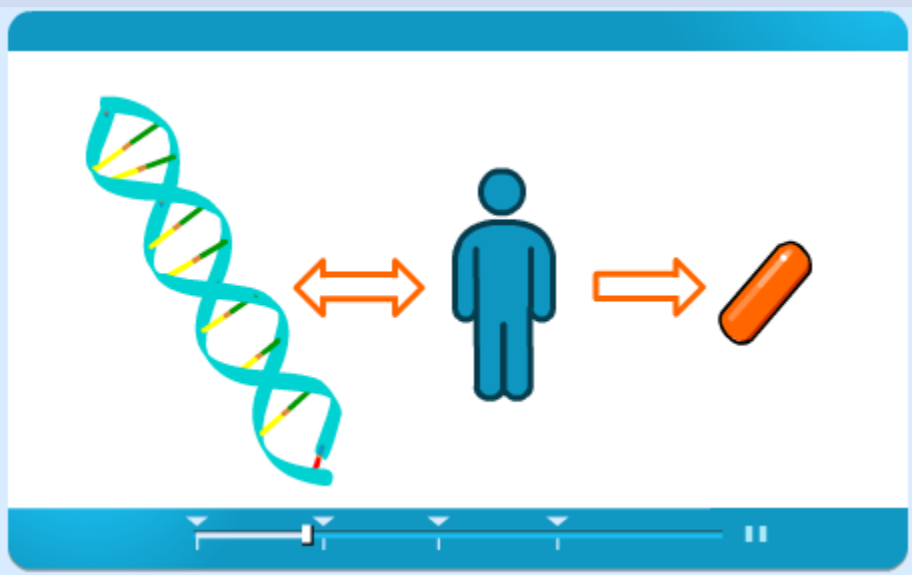
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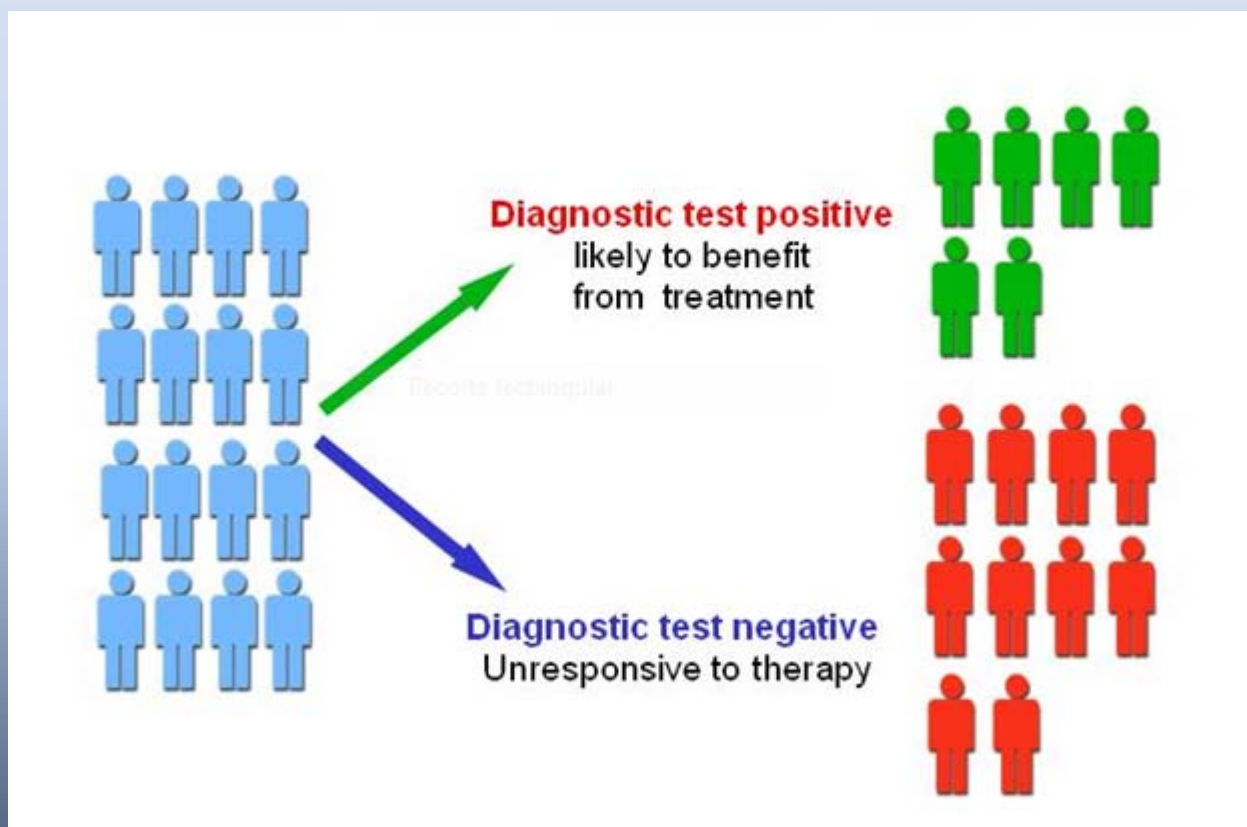
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www.phgfoundation.org/tutorials/pharmacogenomics



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Drugs

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Research Areas

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Table of Valid Genomic Biomarkers in the Context of Approved Drug Labels

Pharmacogenomic information is contained in about ten percent of labels for drugs approved by the FDA. A significant increase of labels containing such information has been observed over the last decade. In order to provide a reference for genomic biomarkers in labels of FDA-approved drug products, we created the table shown below. Genomic biomarkers can play an important role in identifying responders and non-responders, avoiding toxicity and adjusting the dosage of drugs to optimize their efficacy and safety. In the context of drug labels, these genomic biomarkers can be classified on the basis of their specific use, for example:

- Clinical response and differentiation,
- Risk identification,
- Dose selection guidance,
- Susceptibility, resistance and differential disease diagnosis,
- Polymorphic drug targets.

The table portrays a view on valid genomic biomarkers in the context of FDA-approved drug labels. It provides a comprehensive list of these markers and links to pharmacogenomic data, taking into account multiple regulatory contexts in which these biomarkers were approved. Most drug labels in this table provide pharmacogenomic information with no immediate recommendation for a specific action (i.e. genetic testing); however a few labels recommend or require genetic testing thereby specifying the use of these markers for reaching a therapeutic decision.

The table includes:

- Context-specific biomarker (column 1)
- Reference drug label information about the biomarker context within which the drug was approved (column 2 subsection 1)
- Prototypic drug associated with the label information defining the biomarker context (column 2 subsection 2)
- Other drugs in a similar context (column 3)
- Patient population (column 4)

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Proporcionamos servicios de diagnóstico genético a hospitales y clínicas de toda España.



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...para ofrecer tanto a ciudadanos como a profesionales de la salud, información actualizada y rigurosa sobre la Genética y las enfermedades hereditarias, así como una guía para la aplicación a la Medicina de las últimas técnicas de diagnóstico genético, incluyendo servicios de Análisis Genéticos y Consulta de Consejo Genético.

Mientras, comparte con nosotros en



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A 117 personas les gusta esto



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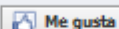


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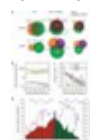


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Genagen <http://www.nature.com/nature/journal/v467/n7319/full/nature09534.html>



A map of human genome variation from population-scale sequencing :
Nature : Nature Publishing Group
www.nature.com

The goal of the 1000 Genomes Project is to provide in-depth information on variation in human genome sequences. In the pilot phase reported here, different strategies for genome-wide sequencing, using high-throughput sequencing platforms, were developed and compared. The resulting data set includes

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Genagen http://www.nlm.nih.gov/medlineplus/spanish/news/fullstory_104869.html



Un estudio halla que las mutaciones genéticas podrían reducir la efectividad de Plavix: MedlinePlus



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